



## PCCB gene

propionyl-CoA carboxylase beta subunit

### Normal Function

The *PCCB* gene provides instructions for making part of an enzyme called propionyl-CoA carboxylase, specifically, the beta subunit of this enzyme. Six beta subunits come together with six alpha subunits (produced from the *PCCA* gene) to form a functioning enzyme.

Propionyl-CoA carboxylase plays a role in the normal processing of proteins. It is responsible for a particular step in the breakdown of several protein building blocks (amino acids) called isoleucine, methionine, threonine, and valine. Propionyl-CoA carboxylase also helps break down certain types of lipids (fats) and cholesterol. First, several chemical reactions convert the amino acids, lipids, or cholesterol to a molecule called propionyl-CoA. Using the B vitamin biotin, propionyl-CoA carboxylase then converts propionyl-CoA to a molecule called methylmalonyl-CoA. Additional enzymes break down methylmalonyl-CoA into other molecules that are used for energy.

### Health Conditions Related to Genetic Changes

#### propionic acidemia

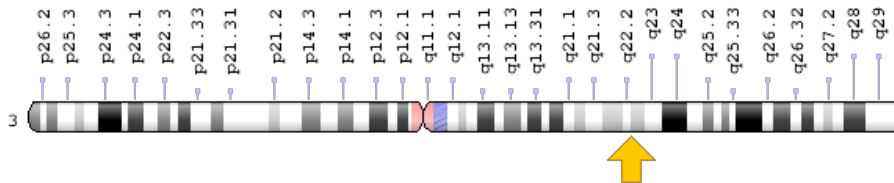
More than 55 mutations in the *PCCB* gene have been identified in people with propionic acidemia. These mutations include changes in single DNA building blocks (nucleotides) and insertions or deletions of genetic material in the *PCCB* gene.

*PCCB* mutations prevent the production of functional propionyl-CoA carboxylase or reduce the enzyme's activity. The altered or missing enzyme prevents certain parts of proteins and lipids from being broken down properly. As a result, propionyl-CoA and other potentially toxic compounds can build up to toxic levels in the body. This buildup damages the brain and nervous system, causing the serious health problems associated with propionic acidemia.

## Chromosomal Location

Cytogenetic Location: 3q22.3, which is the long (q) arm of chromosome 3 at position 22.3

Molecular Location: base pairs 136,250,325 to 136,330,171 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- PCCase beta subunit
- PCCB\_HUMAN
- propanoyl-CoA:carbon dioxide ligase beta subunit
- propionyl CoA carboxylase, beta polypeptide
- propionyl-CoA carboxylase, beta subunit

## Additional Information & Resources

### Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Propionyl-CoA carboxylase deficiency blocks the biotin- and ATP-dependent conversion of propionyl-CoA to methylmalonyl-CoA  
<https://www.ncbi.nlm.nih.gov/books/NBK27933/#A3116>

### GeneReviews

- Propionic Acidemia  
<https://www.ncbi.nlm.nih.gov/books/NBK92946>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PCCB%5BTIAB%5D%29+OR+%28propionyl+Coenzyme+A+carboxylase%5BTIAB%5D%29%29+OR+%28propanoyl-CoA%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### OMIM

- PROPIONYL-CoA CARBOXYLASE, BETA SUBUNIT  
<http://omim.org/entry/232050>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=PCCB%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=8654](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8654)
- Kraus Lab at the University of Colorado Health Sciences Center  
<http://www.ucdenver.edu/academics/colleges/medicalschoo/programs/kraus/Pages/home.aspx>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/5096>
- UniProt  
<http://www.uniprot.org/uniprot/P05166>

### **Sources for This Summary**

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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12757933>

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